

Short Communication on Cancer Genetics

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INTRODUCTION

Cancer is a complex disease that result from the basic process of uncontrolled growth. Cell proliferation results in a mass that invades neighbouring tissues and may metastasize to more distant sites. In order for a normal cell to transform into a cancer cell, genes which regulate cell growth and differentiation must be altered. When normal regulation is altered, uncontrolled growth is initiated and a malignant tumor develop. Cancer is caused by certain genetic changes that occurs at many levels, from gain or loss of entire chromosomes to a mutation affecting a single DNA nucleotide.

Genetic Changes

Genes carry the instructions to make proteins, which do much of the work in our cells. Certain gene changes can cause cells to evade normal growth controls and become cancer. For example, some cancer-causing gene changes increase production of a protein that makes cells grow. Others result in the production of a misshapen, and therefore non-functional, form of a protein that normally repairs cellular damage.

Genetic changes that promote cancer can be inherited from our parents if the changes are present in germ cells. Such changes, called germline changes, are found in every cell of the offspring. There are many different kinds of DNA changes. Some changes affect just one unit of DNA, called a nucleotide. One nucleotide may be replaced by another, or it may be missing entirely. Other changes involve larger stretches of DNA and may include rearrangements, deletions, or duplications of long stretches of DNA. Cancer cells have more genetic changes than normal cells. But each person's cancer has a unique combination of genetic alterations. Some of these changes may be the result of cancer, rather than the cause. As the cancer continues to grow, additional changes will occur.

Hereditary Cancer Syndromes

Inherited genetic mutations play a major role in about 5 to 10

percent of all cancers. Genetic tests for hereditary cancer syndromes can tell whether a person from a family that shows signs of such a syndrome has one of these mutations. Here are examples of genes that can play a role in hereditary cancer syndromes.

- The most commonly mutated gene in all cancers is *TP53*, which produces a protein that suppresses the growth of tumors. In addition, germline mutations in this gene can cause Li-Fraumeni syndrome.
- Inherited mutations in the *BRCA1* and *BRCA2* genes are associated with hereditary breast and ovarian cancer syndrome.
- Another gene that produces a protein that suppresses the growth of tumors is *PTEN*. Mutations in this gene are associated with Cowden syndrome that increases the risk of breast, thyroid, endometrial, and other types of cancer.

Genetic Testing

The testing is done on a small sample of body fluid or tissue-usually blood, saliva, cells from inside the cheek, skin cells, or amniotic fluid.

A positive test result means the laboratory found a specific genetic alteration that is associated with a hereditary cancer syndrome.

A negative test result means that the laboratory did not find the specific alteration that the test was designed to detect. This result is useful when working with a family in which the specific, disease-causing genetic alteration is already known to be present. If the test reveals a genetic change that is common in the general population among people without cancer, the change is called a polymorphism.

Diagnosis and prognosis: Many cancers have a course that is highly variable and therefore difficult to predict. Because cancer genes dictate the aberrant phenotype of cancer cells, genotypic analysis can provide information on the capacity of a given tumor to grow and spread. This information can potentially be used to categorize tumors and to predict their course and responses to therapy.

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Received: November 06, 2020; **Accepted:** November 19, 2020; **Published:** November 26, 2020

Citation: Phaneendra P (2020) Short Communication on Cancer Genetics. J Cell Sci Therapy. 11. 271 doi: 10.35248/2157-7013.20.11.271

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